



spondyloepiphyseal dysplasia congenita

Spondyloepiphyseal dysplasia congenita is an inherited bone growth disorder that results in short stature (dwarfism), skeletal abnormalities, and problems with vision and hearing. This condition affects the bones of the spine (spondylo-) and the ends (epiphyses) of long bones in the arms and legs. Congenita indicates that the condition is present from birth.

People with spondyloepiphyseal dysplasia congenita have short stature from birth, with a very short trunk and neck and shortened limbs. Their hands and feet, however, are usually average-sized. Adult height ranges from 3 feet to just over 4 feet. Abnormal curvature of the spine (kyphoscoliosis and lordosis) becomes more severe during childhood. Instability of the spinal bones (vertebrae) in the neck may increase the risk of spinal cord damage. Other skeletal features include flattened vertebrae (platyspondyly); an abnormality of the hip joint that causes the upper leg bones to turn inward (coxa vara); a foot deformity called a clubfoot; and a broad, barrel-shaped chest. Abnormal development of the chest can cause problems with breathing. Arthritis and decreased joint mobility often develop early in life.

People with spondyloepiphyseal dysplasia congenita have mild changes in their facial features. The cheekbones close to the nose may appear flattened. Some infants are born with an opening in the roof of the mouth (a cleft palate). Severe nearsightedness (high myopia) is common, as are other eye problems that can impair vision. About one quarter of people with this condition have hearing loss.

Frequency

This condition is rare; the exact incidence is unknown. More than 175 cases have been reported in the scientific literature.

Genetic Changes

Spondyloepiphyseal dysplasia congenita is one of a spectrum of skeletal disorders caused by mutations in the *COL2A1* gene. This gene provides instructions for making a protein that forms type II collagen. This type of collagen is found mostly in cartilage and in the clear gel that fills the eyeball (the vitreous). The *COL2A1* gene is essential for the normal development of bones and other tissues that form the body's supportive framework (connective tissues). Mutations in the *COL2A1* gene interfere with the assembly of type II collagen molecules, which prevents bones and other connective tissues from developing properly.

Inheritance Pattern

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- SED congenita
- SED, congenital type
- SEDc
- Spondyloepiphyseal dysplasia, congenital type

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Spondyloepiphyseal dysplasia congenita
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0038015/>

Other Diagnosis and Management Resources

- MedlinePlus Encyclopedia: Clubfoot
<https://medlineplus.gov/ency/article/001228.htm>
- MedlinePlus Encyclopedia: Lordosis
<https://medlineplus.gov/ency/article/003278.htm>
- MedlinePlus Encyclopedia: Retinal Detachment
<https://medlineplus.gov/ency/article/001027.htm>
- MedlinePlus Encyclopedia: Scoliosis
<https://medlineplus.gov/ency/article/001241.htm>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Clubfoot
<https://medlineplus.gov/ency/article/001228.htm>
- Encyclopedia: Lordosis
<https://medlineplus.gov/ency/article/003278.htm>
- Encyclopedia: Retinal Detachment
<https://medlineplus.gov/ency/article/001027.htm>
- Encyclopedia: Scoliosis
<https://medlineplus.gov/ency/article/001241.htm>
- Health Topic: Bone Diseases
<https://medlineplus.gov/bonediseases.html>
- Health Topic: Connective Tissue Disorders
<https://medlineplus.gov/connectivetissuedisorders.html>
- Health Topic: Dwarfism
<https://medlineplus.gov/dwarfism.html>

Genetic and Rare Diseases Information Center

- Spondyloepiphyseal dysplasia congenita
<https://rarediseases.info.nih.gov/diseases/4987/spondyloepiphyseal-dysplasia-congenita>

Additional NIH Resources

- National Institute of Arthritis and Musculoskeletal and Skin Diseases
https://www.niams.nih.gov/Health_Info/Connective_Tissue/

Educational Resources

- Disease InfoSearch: Spondyloepiphyseal dysplasia congenita
<http://www.diseaseinfosearch.org/Spondyloepiphyseal+dysplasia+congenita/6821>
- Johns Hopkins Medicine
http://www.hopkinsmedicine.org/healthlibrary/conditions/adult/orthopaedic_disorders/spondyloepiphyseal_dysplasia_congenita_22,spondyloepiphysealdysplasiacongenita/
- KidsHealth from the Nemours Foundation
<http://kidshealth.org/en/parents/dwarfism.html>
- MalaCards: sed congenita
http://www.malacards.org/card/sed_congenita

- Nemours Children's Health System
<https://www.nemours.org/service/medical/skeletal-dysplasia/spondyloepiphyseal.html?tab=about>
- Orphanet: Spondyloepiphyseal dysplasia and spondyloepimetaphyseal dysplasia
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=253
- Swedish Information Center for Rare Diseases
<http://www.socialstyrelsen.se/rarediseases/congenitalspondyloepiphysealdy>

Patient Support and Advocacy Resources

- Cleft Palate Foundation
<http://www.cleftline.org>
- Human Growth Foundation
<http://hgfound.org/>
- International Skeletal Dysplasia Registry, UCLA
<http://ortho.ucla.edu/isdr>
- Little People of America
<http://www.lpaonline.org>
- National Organization for Rare Disorders (NORD)
<https://rarediseases.org/rare-diseases/spondyloepiphyseal-dysplasia-congenital/>
- Resource List from the University of Kansas Medical Center
<http://www.kumc.edu/gec/support/dwarfism.html>
- The MAGIC Foundation
<https://www.magicfoundation.org/>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22spondyloepiphyseal+dysplasia+congenita%22>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28Osteochondrodysplasias%5BMAJR%5D%29+AND+%28%28spondyloepiphyseal+dysplasia+congenita%5BTIAB%5D%29+OR+%28sedc%5BTIAB%5D%29+OR+%28sed+congenita%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- SPONDYLOEPIPHYSEAL DYSPLASIA CONGENITA
<http://omim.org/entry/183900>

Sources for This Summary

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Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/21924244>

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